I write this on 2nd October 2009 (Jeans for Genes Day), an event that attempts to raise awareness of childhood genetic disorders; hence the timeliness of the review article this month by Knight which provides an up to date review of genetics research for the general physician. We are reminded that significant advances have taken place over the last 20 years that have greatly enhanced understanding of the broad nature of the human genome including its remarkably complex regulation. Furthermore, there is now a greater appreciation of the extent of genetic diversity between individuals. This is a highly complex and technical area that is of immense importance to all practicing clinicians. The review is particularly useful in that it highlights two important implications of genetic variation: susceptibility to disease and the potential for individual tailoring of pharmacological interventions. I enjoyed reading this review for a number of reasons, notwithstanding the fact that while recognising the importance of genetics research, I have always been somewhat intimidated by its technology and jargon. The subject area is demystified to some extent by use of practical examples that have immediate clinical application. A useful glossary is also appended. One key message is that pharmacogenetics is an exiting area with a promise for future development. Having read the review, the general reader will then be in a position to fully appreciate the other paper in this month’s issue that has a genetics theme. A team from Belfast have hypothesised that adult patients with cystic fibrosis (CF) with the G551D mutation had less severe burden of disease than those CF patients who were F508del homozygotes. This was found to be the case and this study clearly illustrates the clinical relevance of current genetic research.

As previously noted, this journal regularly publishes papers that might be roughly classified as “clinical service audits” or in other words, evaluations of how well established protocols are implemented in clinical practice. I have defended this approach on the basis that a protocol that is evidence based and which has consensus professional support is only as good as its execution. I draw your attention to a paper that calls for the agreement for a standard simple management protocol for a relatively common disorder. This month Agarwal and colleagues consider a specific aspect of a phenomenon that will be readily familiar to all physicians involved in acute medical care. They begin their review by reminding us of the high prevalence of alcohol abuse and its impact on health and on health care services. They argue that patients with alcohol addiction are regularly admitted with acute medical or surgical problems. If these patients are abruptly deprived of alcohol, they may develop symptoms of alcohol withdrawal. The early signs and symptoms may be subtle and if overlooked alcohol dependent patients are at risk of rapid physical and mental deterioration. There are varied protocols in place for the recognition and correct management of these patients. The aim of the study was to document variation in the management of acute alcohol withdrawal. Given the pervasiveness of this disorder and the serious nature of its consequences, one would expect to find some degree of consensus in its management. In fact, wide variation was found both in the acute services available and in the way in which the disorder was managed. The authors conclude with a plea for the development of a nationally agreed protocol that would be adopted into routine practice for the acute management of alcohol withdrawal syndrome.

It is now well accepted that the age of onset of type 2 diabetes (T2DM) is falling with more cases commonly reported in the 18–40 year old age group. This observation is closely linked to obesity and severe insulin resistance. There is an emerging body of evidence to suggest that early onset T2DM is not a benign condition. In fact, one large study
found a 14 fold increased risk for myocardial infarction when compared to non diabetic subjects; the increased risk for older T2DM patients was four fold in comparison. Song and Hardisty by means of a cross sectional study have determined the complication rate for T2DM patients who were diagnosed before the age of 40 years. The findings are alarming. It was found that 527 patients in this diagnostic group had high complication rates for cardiovascular disease, retinopathy and neuropathy and furthermore, these were encountered 13–20 years earlier than in older T2DM subjects. It was disappointing to note that early onset T2DM patients had poorer glycaemic control and other indicators of inadequate disease management (untreated hypertension and absence of therapy with statins). The authors have thus identified a group of patients with unmet health care needs and have highlighted an area of diabetes care worthy of further research.

Michael Bannon

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